Note: Performing your original search, ***gaucher disease***, in PubMed will retrieve *2742* citations.

1: Blood Rev. 1998 Jun;12(2):115-33. Related Articles, Links

Enzyme therapy for Gaucher disease: the first 5 years.

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Gaucher disease was first described by Philippe Gaucher in his 1882 medical thesis. Gaucher's original concept was of an unusual epithelioma of the spleen. By the early 1900s, Mandelbaum recognized the systemic nature of the disease. Several children with Gaucher disease were described at the turn of the century, but Rusca described a rapidly progressive fatal neurodegenerative type of disease, i.e. type 2, in the 1920s. The 'juvenile' form (type 3) of the disease was described in Sweden in the 1950s. In 1965, the deficient enzyme, acid beta-glucosidase, was discovered and the lysosomal nature of the disease was elucidated. Currently, three variants of Gaucher disease have been defined clinically and are distinguished by the presence and severity of neuronopathic involvement (Table 1). Each of these clinical types has substantial phenotypic variation, but types 1 and 3 have significantly heterogeneous rates of disease progression and degrees of visceral organs involvement. The neuronopathic involvement in type 3 also has substantial variation in the age of onset and disease progression even within relatively isolated communities. An extensive review of the clinical and pathologic involvement by Gaucher disease is available.

Publication Types:
- Review

MeSH Terms:
- Animals
- Diphosphonates/therapeutic use
- Dose-Response Relationship, Drug

- Gaucher Disease/drug therapy*
- Gaucher Disease/pathology
- Gaucher Disease/physiopathology
- Glucosylceramidase/pharmacokinetics
- Glucosylceramidase/therapeutic use*
- Humans
- Puberty, Delayed/chemically induced
- Recombinant Proteins/therapeutic use
- Research Support, Non-U.S. Gov't
- Research Support, U.S. Gov't, P.H.S.

Substances:

- Diphosphonates
- Recombinant Proteins
- alglucerase
- imiglucerase
- Glucosylceramidase

Grant Support:

- DK 36729/DK/NIDDK
- NS 34071/NS/NINDS
- NS 36681/NS/NINDS
- etc.

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